



## CHD2 gene

chromodomain helicase DNA binding protein 2

### Normal Function

The *CHD2* gene provides instructions for making a protein called chromodomain DNA helicase protein 2. This protein is found in cells throughout the body and regulates gene activity (expression) through a process known as chromatin remodeling. Chromatin is the complex of DNA and proteins that packages DNA into chromosomes. The structure of chromatin can be changed (remodeled) to alter how tightly DNA is packaged. When DNA is tightly packed, gene expression is lower than when DNA is loosely packed.

### Health Conditions Related to Genetic Changes

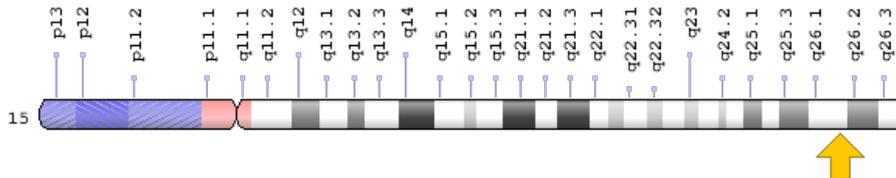
#### CHD2 myoclonic encephalopathy

At least 30 mutations in the *CHD2* gene have been found to cause *CHD2* myoclonic encephalopathy, a condition characterized by recurrent seizures (epilepsy), abnormal brain function (encephalopathy), and intellectual disability beginning in childhood. About half of these mutations delete pieces of DNA from the *CHD2* gene. These and other *CHD2* gene mutations either prevent the production of any chromodomain DNA helicase protein 2 or lead to the production of a nonfunctional version of the protein. As a result, chromatin remodeling and gene expression normally regulated by the chromodomain DNA helicase protein 2 are disrupted. It is unclear why *CHD2* gene mutations seem to only affect nerve cells in the brain or how they lead to the signs and symptoms of *CHD2* myoclonic encephalopathy.

## Chromosomal Location

Cytogenetic Location: 15q26.1, which is the long (q) arm of chromosome 15 at position 26.1

Molecular Location: base pairs 92,900,321 to 93,028,007 on chromosome 15 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- ATP-dependent helicase CHD2
- CHD-2
- EEOC
- FLJ38614

## Additional Information & Resources

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Chromosomal DNA and Its Packaging in the Chromatin Fiber  
<https://www.ncbi.nlm.nih.gov/books/NBK26834/>

### GeneReviews

- CHD2-Related Neurodevelopmental Disorders  
<https://www.ncbi.nlm.nih.gov/books/NBK333201>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CHD2%5BTIAB%5D%29+OR+%28chromodomain+helicase+DNA+binding+protein+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

## OMIM

- CHROMODOMAIN HELICASE DNA-BINDING PROTEIN 2  
<http://omim.org/entry/602119>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_CHD2.html](http://atlasgeneticsoncology.org/Genes/GC_CHD2.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=CHD2%5Bgene%5D>
- HGNC Gene Family: DNA helicases  
<http://www.genenames.org/cgi-bin/genefamilies/set/1167>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=1917](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=1917)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/1106>
- UniProt  
<http://www.uniprot.org/uniprot/O14647>

## **Sources for This Summary**

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*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/26262932>
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<https://ghr.nlm.nih.gov/gene/CHD2>

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